



PROPOSED AMENDED
FIGURES 4-6

09/707,919

4/18

http://www.ncbi.nlm.nih.gov/SNP/snp_retrieve.cgi?subsnp_id=8697



NCBI

SNP Details

SNP Details

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SNP:

Handle|local_snp_id: FGU-CBT | SKB.2K.1.1

NCBI Assay Id(ss#): 869704

Reference SNP Id(rs#): 695871

STS Information: Not submitted

From SNP Database:

Submitter Handle: FGU-CBT
 Submitter Batch ID: SKB.2K.1
 Release Date: Aug 2 2000 2:53PM
 Molecular type: Genomic
 No. of Chromosomes sampled: 215
 Synonym defined:
 Organism: Homo sapiens
 Population: INDRP
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

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NCBI Assay ID: 869704
 Submitter SNP ID: SKB.2K.1.1
 Synonyms:
 LOCUSID: 6311
 Submitter STS ID: not available
 STS Accession: U70323
 GenBank Accession: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene
 Gene Name: 459
 Length:

Flanking Sequence Information:

5' Assay: CTCGCCCTCA GACTGTTTGT GTAGCAACGG CAACGGCGGC GCGCGGTTTC GGCCCGGCTC
 CCGCGGGCTC CTTGGTCTCG GCGGGCCTCC CCGCCCTTC GTGCTC (SEQ ID NO: 14)

Observed: G/C

3' Assay: TCCTTCTCCC CCTCGCCAGC CCGGGCGCCC CTCGGGCGGC GCCAACCCGC GCCTCCCCGC
 TCGGCGCCCG TCGGTCCCCG CCGCGTTCGG CGGTCTCCTT GCGCGGCGCC GCTCCCGGCT
 GTCCCCGCCC GCGGTGGAG CCGGTGTATG GCGCCCTCAC CATGTGCT (SEQ ID NO: 15)

3' Flank: GAAGCCCCAG CAGCAGCAGC AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA
 GCAGCAGCAG CAGCCGCGCG CCGCGGCTGC CAATGTCCGC AAGCCCGCGG GCAGCGGCCT
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 CAC (SEQ ID NO: 16)

Allele Frequency Information:

POP Batch Id: SKB.2K.1:

Submitter Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT | INDRP
 No. of Chromosomes Sampled: 215

Details

Allele: C = 0.293 / G = 0.707

Fig.4(Cont.)

9/26/00 6:50 PM



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Reference SNP Record

NCBI SNP ID: rs695871

General

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NCBI Resource Links

GenBank: [U70323](#)

LocusLink: no link established

Integrated Maps: *under construction*

Submitter records for this ID

Search

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Assay ID Handle | Local Submitter ID

[ss869704](#) FGU-CBT|SKB.2K.1.1

Release Date

Aug 2 2000 2:53PM

Variation Summary

Assay sample size (number of chromosomes) : 430
 Population data sample size (number of chromosomes) :
 Total number of populations with frequency data: 1
 Total number of individuals with genotype data: 0
 Average estimated heterozygosity: 0.414
 Average Allele Frequency:

C:	0.293
G:	0.707

Validation Summary

Marker displays Mendelian segregation: UNKNOWN
 PCR results confirmed in multiple reactions: YES
 Homozygotes detected in individual genotype data: UNKNOWN
 Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg
 Insufficient data to compute individual x genotype consistency measures
 Validation status: *under construction*

ter Contact Details

http://www.ncbi.nlm.nih.gov/SNP/snp_view...ype=contact&handle=FGU-CBT&batch_id=2


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 institution: Centre for Biochemical Technology (CSIR)
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.



SNP Population Details - 558

General

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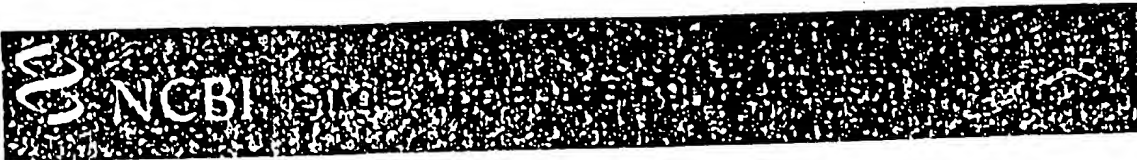
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Submitter Population ID: INDPOP
Population Text:

Continent: Asia
Nation: India

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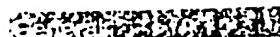
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Method

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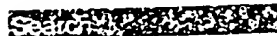

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[Blast SNP](#)
[Submission Form](#)

Submitter Method Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Method Text:

(SEQ ID NO: 1)

(SEQ ID NO: 2)

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCCGCCCTCAGACTGTTTGGTAG 3') and SCA2-RP3 (5' GTGGCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 µl reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl₂, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

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Publication

SNP Publication Details

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Submitter Handle: FGU-CBT

pmid:

MEDUID:

TITLE:

Single Nucleotide Polymorphism in SCA2 Gene.

AUTHOR:

CHOUDHRY, S.; BRAHMACHARI, S.K.

JOURNAL:

VOLUME:

SUPPL:

ISSUE:

I_SUPPL:

PAGES:

YEAR: 2000

STATUS: 1- unpulished

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Search PubMed by author:

CHOUDHRY, S.; BRAHMACHARI, S.K.

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SNP:

Handle|local_snp_id: FGU-CBT | SKB.2K.1.2
 NCBI Assay Id(ss#): 869705
 Reference SNP Id(rs#): **696872**

STS Information: Not submitted

From SNP Database:

Submitter Handle: **FGU-CBT**
 Submitter Batch ID: **SKB.2K.1**
 Release Date: **Aug 2 2000 2:53PM**
 Molecular type: **Genomic**
 No. of Chromosomes sampled: **215**
 Synonym defined:
 Organism: **Homo sapiens**
 Population: **INDRCP**
 Submitter Method ID: **SCA2-SNP**
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

[View citation details](#)

NCBI Assay ID: 869705
 Submitter SNP ID: SKB.2K.1.2
 Synonyms:
 LOCUSID: **6311**
 Submitter STS ID:
 STS Accession: not available
 GenBank Accession: **U70323**
 Gene Name: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene
 Length: 459

Flanking Sequence Information:

5' Assay: CTCGCCCTCA GACTGTTTGT GTAGCAACGG CAACGGCGGC GGCGCGTTTC GGCCCGGCTC
 CCGGCGGGTC CTGGTCTCG GCGGGCTCC CCGCCCTTC GTGTCGTCC TTCTCCCCCT
 CGCCAGCCCG GGCGCCCTC CGGCCGCGC AACCCGCGC TCCCCGCTC GCGCCCG (SEQ ID NO: 17)

Observed: T/C

3' Assay: GCGTCCCCGC CGCGTTCGG CGTCTCCTTG GCGCGCCCG CTCCCGGCTG TCCCCGCCCC
 GCGTGCGAGC CGGTGTATGG GCCCCCACC ATGTGCT (SEQ ID NO: 18)

3' Flank: GAAGCCCCAG CAGCAGCAG AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA
 GCAGCAGCAG CAGCCGCGCC CCGCGGCTGC CAATGTCCGC AAGCCCGCGC GCAGCGGCCT
 TCTAGCGTCG CCCGCGCGCG CGCCTTCGCC GTCTCTGTCC TCGGTCCTCT CGTCTCTGGC
 CAC (SEQ ID NO: 16)

Allele Frequency Information:

POP Batch Id: SKB.2K.1:

Submitter Handle: **FGU-CBT**
 Submitter Method ID: **SCA2-SNP**
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT|INDRCP
 No. of Chromosomes Sampled: 215
 Allele: C = 0.293 / T = 0.707



RefSNP Record

Reference SNP Record

NCBI SNP ID: rs695872

NCBI Resource Links

GenBank: [U70323](#)

LocusLink: no link established

Integrated Maps: *under construction*

Submitter records for this ID:

Assay ID	Handle Local Submitter ID	Release Date
ss869705	FGU-C8T SKB.2K.1.2	Aug 2 2000 2:53PM

Variation Summary:

Assay sample size (number of chromosomes) : 430
 Population data sample size (number of chromosomes) :
 Total number of populations with frequency data: 1
 Total number of individuals with genotype data: 0
 Average estimated heterozygosity: 0.414
 Average Allele Frequency:

C:	0.293
T:	0.707

Validation Summary:

Marker displays Mendelian segregation: UNKNOWN
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 Homozygotes detected in individual genotype data: UNKNOWN
 Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg
 Insufficient data to compute individual x genotype consistency measures
 Validation status: *under construction*

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Publication

SNP Publication Details

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Submitter Handle: FGU-CBT

pmid:

MEDID:

TITLE:

Single Nucleotide Polymorphism in SCA2 Gene.

AUTHOR:

CHOUDHRY,S.; BRAHMACHARI,S.K.

JOURNAL:

VOLUME:

SUPPL:

ISSUE:

I_SUPPL:

PAGES:

YEAR:

2000

STATUS:

1- unpublished

Search PubMed by author:

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Population

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Submitter Population Handle: FGU-CBT

Submitter Population ID: INDPOP

Population Text:

Continent: Asia

Nation: India

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Method

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Submitter Method Handle: FGU-CBT

Submitter Method ID: SCA2-SNP

Method Text:

(SEQ ID NO:1)

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCGCCCTCAGACTGTTTGGTAG 3') and SCA2-RP3 (5' GTCGCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 µl reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl₂, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

(SEQ ID NO:2)

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Submitter

Submitter Contact Details

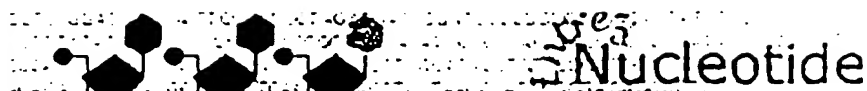
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 email: skb@cbt.res.in
 lab: Functional Genomics Unit
 institution: Centre for Biochemical Technology (CSIR),
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.



Search for

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☐ 1: GI = "1679683" [GenBank] Human ataxin-2 (SCA2) mRNA.... PubMed, Protein, Related Sequences, Taxonomy, OMIM, LinkOut

LOCUS HSU70323 4481 bp mRNA PRI 20-NOV-1996
 DEFINITION Human ataxin-2 (SCA2) mRNA, complete cds.
 ACCESSION U70323
 VERSION U70323.1 GI:1679683
 KEYWORDS
 SOURCE human.
 ORGANISM Homo sapiens
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (bases 1 to 4481)
 AUTHORS Pulst,S.-M., Nechiporuk,A., Nechiporuk,T., Gispert,S., Chen,X.-N.,
 Lopes-Cendes,I., Pearlman,S., Starkman,S., Orozco-Diaz,G.,
 Lunkes,A., DeJong,P., Rouleau,G.A., Auburger,G., Korenberg,J.R.,
 Figueroa,C. and Sahba,S.
 TITLE Moderate expansion of a normally biallelic trinucleotide repeat in
 spinocerebellar ataxia type 2
 JOURNAL Nature Genet. 14 (3), 269-276 (1996)
 MEDLINE 97051920
 REFERENCE 2 (bases 1 to 4481)
 AUTHORS Pulst,S.-M.
 TITLE Direct Submission
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 Los Angeles, CA 90048, USA
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Figure : 6

Wednesday, September 27, 2000

NCBI Sequence Viewer

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ORIGIN

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2221  catagaccac ggtctccccc acagaacagt attggaataa ccccagtggt gccagtctct
2281  gcttctcccc aagctgttat tattccaact gaagctgttg ccatgcctat tccagctgca
2341  tctcctacgc ctgctagttc tgcctcgaac agagctgtta ccccttctag tgaggctaaa
2401  gattccaggg ttcaagatca gaggcagaac tctcctcgag ggaataaaga aaatatataa
2461  cccaatgaaa catcacatag cttctcaaaa gctgaanaaa aaggtatctc accagtgtgt
2521  tctgaacata gaaaacagat tgatgattta aagaaattta agaattgatt taggttacag
2581  ccaagtctta cttctgaatc tatggatcaa ctactaaca aaaaatagaa gggagaaaaa
2641  tcaagagatt tctcgaagaa caaaattgaa ccaagtgtca aggtattctt cattgaaaaa
2701  agcagcagca actgtaccag tggcagcagc aagccgaata gccccagcat ttccccctca
2761  atacttagta acacggagca caagagggga cctgaggtca ctcccaagg ggttcagact
2821  tccagccagc catgtaaaac agagaagac gataaggaag agaagaaga cgcagctgag
2881  caagttagga aatcaacatt gaatcccaat gcaaaaggag tcaacccagc ttctctctct
2941  cagccaaaagc cttctactac cccaacttca cctcggcctc aagcacaacc tagcccatct
3001  atggtgtgtc atcaaacgcc aactccagtt tatactcagc ctgtttgttt tgcacaaaat
3061  atgatgtatc cagtcccggt gagcccgagg gtgcaacctt tatacccaat acctatgacg
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3181  caagaccagc atcatcagag tggcatgatg caccagcggt cagcagcggg cccaccgatt
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3361  cctgtaatac agggtaatgc tagaatgatg gcaccaccaa cacacgcccc gcttggttta
3421  gtatctctct cagcaactca gtacggggct catgagcaga cgtatcgatg gtatgcatgt
3481  cccaaattac catacaacaa ggagacaagc ccttcttctt actttgccaat tccacgggc
3541  tcccttgcct agcagtatgc gcaccctaac gtaaccttgc acccacatac tccacacctc
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3721  cagcagcagt cagccattta ccacgcgggg ctbgcgccaa ctccacctc catgacacct
3781  gcctccaaca cgcagtcgcc acagaatagt ttcccagcag caaacagac tgtcttttag
3841  atccatctct ctacagttca gccggtgtat accaaccac cccacatggc ccacgtacct
3901  caggtcatgt tacagttagc aatggttctt tctcatccaa ctgcccattg gccaatgatg
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4261  tgggaagatac ttggaccggc tagaggtcat taggaacttg ggggtctatt cataattcca
4321  tatgctgttt cagagtcccc caggtacccc agctctgctt gccgaactg gaagtatttt
4381  attttttaat aacccttgaa agtcatgaac acatcagcta gcaaaagaag taacaagagt
4441  gattcttgct gctattactg ctaaaaaaa aaaaaaaa a

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(SEQ ID NO: 20)

file:///C:/Users/.../Documents/.../Fig.6(Cont.)

Fig.6(Cont.)